



## FRAS1 gene

Fraser extracellular matrix complex subunit 1

### Normal Function

The *FRAS1* gene provides instructions for making a protein that is part of a group of proteins called the FRAS/FREM complex. This complex is found in basement membranes, which are thin, sheet-like structures that separate and support cells in many tissues. The FRAS/FREM complex is particularly important during development before birth. One of its roles is to anchor the top layer of skin by connecting the basement membrane of the top layer to the layer of skin below. The FRAS/FREM complex is also involved in the proper development of certain other organs and tissues, including the kidneys, although the mechanism is unclear.

### Health Conditions Related to Genetic Changes

#### Fraser syndrome

At least 24 mutations in the *FRAS1* gene cause a condition called Fraser syndrome; *FRAS1* gene mutations account for about half of cases of this condition. Fraser syndrome affects development before birth and is characterized by eyes that are completely covered by skin (cryptophthalmos), fusion of the skin between the fingers and toes (cutaneous syndactyly), and abnormalities of the kidneys and other organs and tissues.

Most mutations in the *FRAS1* gene involved in Fraser syndrome lead to production of an abnormal protein with little or no function. The abnormal protein is likely broken down quickly, preventing formation of the FRAS/FREM complex. Lack of the FRAS/FREM complex in the basement membrane of the skin leads to detachment of the top layer, causing blisters to form during development. These blisters likely prevent the proper formation of certain structures before birth, leading to cryptophthalmos and cutaneous syndactyly. It is unknown how lack of the FRAS/FREM complex leads to kidney abnormalities and other problems in Fraser syndrome.

#### other disorders

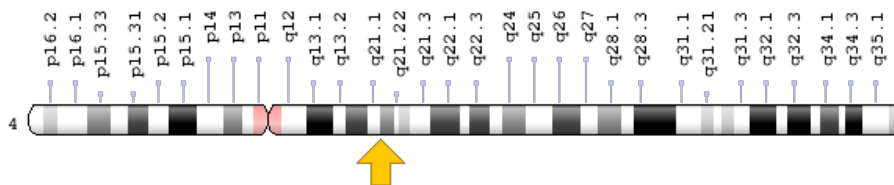
Mutations in the *FRAS1* gene have also been found in people with abnormalities of the kidneys and urinary tract but no other signs and symptoms of Fraser syndrome (described above). Such abnormalities are grouped together as congenital anomalies of the kidney and urinary tract (CAKUT). A common abnormality in people with *FRAS1* gene mutations is the absence of one of the kidneys (a condition called renal agenesis). The *FRAS1* gene mutations involved in CAKUT typically change

single protein building blocks (amino acids) in the FRAS1 protein. Researchers speculate that the effects of these mutations are milder than those of mutations that cause Fraser syndrome; some FRAS1 protein function may still remain. How these gene mutations affect the FRAS/FREM complex or lead to renal agenesis and other CAKUT is unknown.

### Chromosomal Location

Cytogenetic Location: 4q21.21, which is the long (q) arm of chromosome 4 at position 21.21

Molecular Location: base pairs 78,057,323 to 78,544,269 on chromosome 4 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

### Other Names for This Gene

- extracellular matrix protein FRAS1
- FLJ14927
- FLJ22031
- Fraser syndrome 1
- KIAA1500

### Additional Information & Resources

#### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28FRAS1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D>

## OMIM

- CONGENITAL ANOMALIES OF KIDNEY AND URINARY TRACT 1, SUSCEPTIBILITY TO  
<http://omim.org/entry/610805>
- FRAS1 GENE  
<http://omim.org/entry/607830>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=FRAS1%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=19185](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=19185)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/80144>
- UniProt  
<http://www.uniprot.org/uniprot/Q86XX4>

## **Sources for This Summary**

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